Headstart Booklet - 2015 Year 10 Science

**Genetics**

Name\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Form\_\_\_\_\_\_\_\_\_

Teacher \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

By the end of this unit you will have an understanding of:

* Genetic terms
* Patterns of inheritance
* Genetic disorders

**Task 1: Comparing Traits**

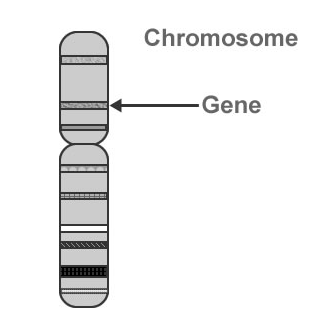
1. Study the different characteristics shown below and determine which of these traits you show – circle the trait you have. Tally the class totals. Were the dominant traits the most common? Yes/No

|  |  |  |  |
| --- | --- | --- | --- |
| **Tongue Rolling**  (rolling is dominant) | http://www.biologyjunction.com/images/images23.jpgCan roll tongue  http://www.biologyjunction.com/images/images22.jpgCan’t roll tongue | **Widow’s peak**  (peak is dominant) | Widow’s peak  Straight hairline |
| **Ear lobe attachment**  (free is dominant) | Attached Non-attached | **Hitchhiker’s thumb**  (straight is dominant) | http://www.biologyjunction.com/images/clip0030.jpg |
| **Bent little finger**  (straight is dominant) | http://www.biblenews1.com/hands/pictures/finger4bent.gif | **Dimples**  (dimples are dominant) | http://www.biologyjunction.com/images/Dimples20Deluxe1.jpghttp://www.biologyjunction.com/images/images44.jpg  Dimples No dimples |
| **Mid-digit hair**  (hair is dominant) | http://faculty.stcc.edu/BIOL102/images/labs/genetics/hairfinger.gif | **Hand clasping**  (left thumb over is dominant) | http://www.biologyjunction.com/images/clip0032.jpg  Left on top Right on top |
| **Cleft chin**  (cleft is dominant) | Cleft chin Non-cleft chin | **Darwin’s point**  (present is dominant) |  |

**Task 2: Dominant/Recessive – what does this mean?**

**Task 3: Journey into DNA**

Before watching the animation [http://www.pbs.org/wgbh/nova/genome/dna.html#](http://www.pbs.org/wgbh/nova/genome/dna.html) write everything you know about **DNA, chromosomes, genes, alleles, phenotype and genotype** in the box below

After listening to your teacher’s summary of DNA, chromosomes, genes, alleles, phenotype and genotype, draw another chromosome in the picture below and add labels to show your understanding of alleles

**Task 4: Homozygous/Heterozygous – what does this mean?**

**Task 5: Quick review of genetic terms used so far**

Match the following terms with their definitions by writing the corresponding term in the third column. The first one has been done for you

|  |  |  |
| --- | --- | --- |
| **Terms** | **Definitions** | **Matching term** |
| Allele for the dominant trait | 1. A specific sequence of DNA that codes for a particular trait (e.g. eye colour) | gene |
| Allele for the recessive trait | 1. Having two copies of the same allele (e.g. AA or aa) |  |
| Gregor Mendel | 1. Different forms of the same gene (e.g. you may have one for blue eyes and one for brown eyes) |  |
| Chromosome | 1. The allele that masks the effect of the recessive trait. An individual only needs one copy of this allele for it to be expressed (see in the phenotype) |  |
| Gene | 1. The ‘father’ of genetics. His experiments on pea plants are responsible for much of our knowledge of genetics. |  |
| Allele | 1. Having two different alleles (e.g. Aa) |  |
| Homozygous or pure-breeding | 1. Structures within the nucleus of cells that are made up of DNA |  |
| Heterozygous or hybrid | 1. When a trait is carried on an X or Y chromosome |  |
| Genotype | 1. The combination of genes an individual has (e.g. Bb) |  |
| Phenotype | 1. A ‘map’ of all the genetic material in humans |  |
| Pedigree | 1. A chemical that makes up chromosomes and determines the particular traits we have (i.e. what we look like) |  |
| Sex-linked inheritance | 1. A diagram used to trace the inheritance of traits through a family. |  |
| DNA | 1. The allele that is masked by the dominant trait. An individual needs two copies of this allele for it to be expressed (seen in the phenotype) |  |
| Gene expression | 1. The physical characteristics of an individual (e.g. brown hair) |  |
| Human genome | 1. The appearance of characteristics in an organism that are coded for by genes |  |

**Task 6: Pedigrees – what do you know**

Now watch the YouTube clips and discover more about pedigrees: <http://www.youtube.com/watch?v=Ir1t9awmUl4>

**Task 7: Pedigree analysis – complete the questions 1-9 (page 5 & 6)**

**Introduction**

As we have discovered from the exercise on page 1 your class illustrates many similarities between unrelated individuals – we share hair, eyes, nose, arms, legs, and many other characteristics. Of course, there are many differences in the form of these characteristics between members of the group. For example, hair can be blonde, brown, red or black; it can be straight, wavy or curly; it may be fine in some individuals and coarse in others. There are enormous variations in eye colour. But look within one single family – there are more similarities between related individuals within a family than there are between unrelated members in your class. For example, fair skin and reddish hair are likely to be shared by individuals within one family. These characteristics are also likely to be shared by other generations within the family. Why is this? Family trees or pedigrees provide a useful way of analysing information that is inherited from one generation to the next within a family. Analysis of pedigrees gives us a clue about the way characteristics are inherited, and their pattern or mode of inheritance. They can also be used to determine the genetic make-up, or genotype of an individual, and to predict the chances of children of having particular features.

**Purpose**

To analyse selected pedigrees to determine the mode of inherited of genetic traits

To predict the possible outcomes in children born of particular partnerships in relation to inherited diseases.

To construct pedigrees from family histories in order to determine modes of inheritance.

**Procedure**

Carefully read the foundation ideas in the background notes below. This will familiarise you with the symbols used in pedigree analysis. The information in the table below outlines the key features that distinguish the modes of inheritance for different phenotypic characteristics. Use this information to help you answer the questions.

**Background**

In pedigree analysis, symbols are used to provide specific information about individuals in a clear and simple way. Use the following legend as a guide to interpreting the pedigrees in this activity.

Female Affected female

Male Affected male

Non-identical twins Identical twins

|  |  |
| --- | --- |
| **Pattern of Inheritance** | **Key Features** |
| Autosomal dominant | Affected individuals must carry at least one dominant allele  Unaffected parents will not produce affected offspring (unless a new mutation occurs)  Any individual with the trait must have at least one parent with the trait  Two affected parents may produce non-affected child |
| Autosomal recessive | Affected individuals are homozygous recessive  Unaffected parents can produce affected offspring |
| X-linked dominant | Affected males pass trait to all of their daughters and none of their sons  Any individual with the trait must have at least one parent with the trait |
| X-linked recessive | Affected females produce only affected sons  Expect half the sons of unaffected female carriers to be affected |

**Pedigree Analysis**

*Scenario 1*

The pedigrees below show the inheritance pattern of earlobe shape in two different families.. ‘Free lobes’ are dominant to ‘attached lobes’, which are recessive. The gene responsible for earlobe shape has two alternative alleles represented by E (free lobes) and e (attached lobes).

a. b.

1. Assign genotypes to all individuals in pedigree a.
2. Describe the pattern or mode of inheritance for earlobe shape in humans
3. Examine pedigree b. Try to assign genotypes to all individuals. Why is it difficult to do this with confidence?
4. Write in the possible genotypes for all those that are inconclusive in pedigree b.

*Scenario 2*

Known family histories are also useful to geneticists in establishing the mode of inheritance for particular genetic diseases. Pedigree analysis for families that show such diseases is also important so that genetic counseling can be provided to families about the likelihood of future children being affected or carrying the allele in question. The pedigrees below illustrate the inheritance of Huntington’s disease in two unrelated families. Huntington’s disease is a neurological disorder that leads to gradual, permanent deterioration of nerve and muscle control with eventual complete dependence on care. Death results after some years. The onset of the symptoms does not occur until at least mid to late thirties.

B

A

1. Assign genotypes to each person in both pedigrees.
2. Name the mode of inheritance for Huntington’s disease. Explain your choice.
3. Individuals A and B are engaged to be married. Both individuals are keen to raise a family. What are the chances of any children from this union developing Huntington’s disease? Show your working.

*Scenario 3*

People with galactosaemia are unable to digest milk sugar (galactose).

[Type a quote from the document or the summary of an interesting point. You can position the text box anywhere in the document. Use the Drawing Tools tab to change the formatting of the pull quote text box.]

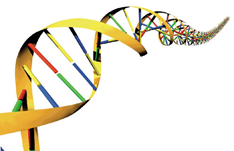
1. From the evidence of the pedigrees above, suggest which pedigree shows beyond doubt that galactosaemia is inherited as an autosomal recessive condition. Explain your reasoning.

*Scenario 4*

Red-green colour blindness is a relatively common condition, inherited as an X-linked recessive trait. The pedigrees below show three families in which this condition occurs.

1. Which of the three pedigrees best establishes the mode of inheritance for this trait? Explain your reasoning.

**Task 8: Genetic disorders – what do you know about Tay Sachs Disease, Duchenne’s Muscular Dystrophy, Haemophilia, Cystic Fibrosis and Huntington’s Disease?**

******Task 9: Genetics Counselling Task – the research**

**Introduction**

In this task you will work in groups of 3 to research a genetic disease. The information you collect and materials you develop will be used by your group to counsel a couple who want to have children, but are worried that their child may inherit this disease. You must consider all of the information a couple may want or need to know to help them make a decision on whether or not to have offspring.

To assist you in giving the consultation you are required to develop support materials. They will be used both to prompt you during the consultation and to help explain things to the client. These materials should take a variety of forms such as written, visual, auditory……can you think of anything else?

**What will happen on presentation day?**

* Half the groups in the class will set up a consultation room (these are the consultation groups)
* The remaining groups (the client groups) will be given a scenario. This scenario will direct them to a specific consultation area.
* Two people in the client group will be counselled, while the third films the consultation.
* The consultation group must help the couple based on the specific scenario **and** the information they have researched. All group members must contribute equally to the consultation and will be given **10 minutes** to complete their consultation.
* The clients should remember to ask questions when they do not understand to give the consultation group an opportunity to re-explain a concept.
* The consultation groups and client groups will swap and repeat the process above.
* Each group will evaluate the group they were counselled by using a rubric provided to them after all groups have presented.

**How long will this task take?**

The full task will take 5 periods. The following table shows what you will be expected to do in each of these periods. A planning sheet will also be provided to assist you.

|  |  |
| --- | --- |
| **Period** | **Work to complete** |
| 1 | Research & develop support materials – to be finished in Headstart  bibliography must be listed on page 13 of this booklet |
| 2 |
| 3 |
| 4 | Complete consultation & video (completed in 2014) |
| 5 | Watch videos & complete peer assessment (completed in 2014) |

**How will we be assessed?**

You will be assessed in 3 ways:

* *Teacher assessment:* video of the consultation.
* *Teacher assessment:* choose ONE piece of your support material for your teacher to look at in depth and submit this after the consultation, as well as any additional materials you create during the consultation to assist the understanding of your clients.
* *Peer assessment:* your clients will complete an evaluation form of your consultation.

**Questions?**

**Planning Sheet**

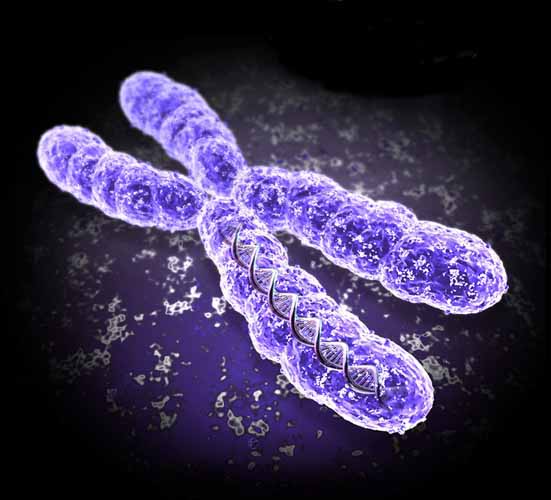
To perform your duties as a genetic counsellor effectively, you will first need to determine what a genetic counsellor actually does and what sort of information they provide to their clients. What would **YOU** want to know if you wanted to have a child who could potentially inherit a genetic disease? Remember your clients are not geneticists.

Research the role of a genetic counsellor and brainstorm what information you will need to find:

Role of a genetic counsellor:

What information will we need to research about our disease and how it is inherited?

Consider **ALL** the information we have covered so far in genetics. How can you apply this to your genetic counselling session? How will you work through a specific scenario and inform the parent both about the specific disease and whether or not their child will inherit it. What will you need to know and need to be able to do? Write your ideas here:



What materials do you need to create to help you complete the consultation? Remember this will be handed in for assessment. Will this involve audio, written material, visual material.....you MUST let your teacher know what you require so it can be booked in time to be used in your planning lessons.

On consultation day what materials might be useful to help you work through the specific scenario that you will be given? Will you need anything else other than your support materials? Will you use any props?

Who will be responsible for what? Divide your tasks evenly to ensure your group works productively.

|  |  |
| --- | --- |
| **Group Member** | **Responsible For** |
|  |  |
|  |  |
|  |  |
|  |  |

**Task 10: Peer Evaluation Sheet – to be used next year**

**Genetic counselling group (group being assessed): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**Client group (group assessing): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

Use the following rubric to rate how well your genetic counsellors covered the following criteria:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Criteria** |  |  |  |  |
| *The disease and its symptoms* | The group gave an overview of the disease and described in detail the symptoms that an individual with the disease may experience. | The group gave an overview of the disease and described some symptoms that an individual with the disease may experience. | The group didn’t really explain the disease and only mentioned a few symptoms. | The consultation did not cover this section. |
| *Life expectancy and quality of life* | The group stated how long individuals with the disease typically live and explained in detail the quality of life expected. | The group stated how long individuals typically live and briefly explained the quality of life expected. | The group stated how long individuals typically live but did not mention quality of life. | The consultation did not cover this section. |
| *Chances of the child inheriting the disease* | The group explained the chance of a child of the couple inheriting the disease (either as a percentage, ratio or fraction) and showed the couple precisely how this was determined using Punnett squares **AND** pedigrees. | The group stated the chance of a child of the couple inheriting the disease (either as a percentage, ratio or fraction) and showed the couple how this was determined using Punnett squares **OR** a pedigree. | The group stated the chance of child of the couple inheriting the disease (either as a percentage, ratio or fraction) but did not show the couple how this was determined. | The consultation did not cover this section. |
| *Treatment and management of the disease if the child should inherit it* | The group explained the different options for treatment or management in detail. | The group explained some options for treatment or management of the disease. | The group briefly explained one treatment or management option. | The consultation did not cover this section. |
| *Impact on parents and* | The group discussed in detail the impact the disease is likely to have on the parents in a variety of areas including socially, emotionally and financially | The group briefly discussed the impact the disease is likely to have on the parents in a variety of areas including socially, emotionally and financially | The group mentioned the impact the disease is likely to have on the parents | The consultation did not cover this section. |
| *Where parents can get support* | The group provided a number of places the couple can get support and what these places can offer | The group provided one place the couple can get support and what this place can offer | The group provided places to get support but did not state what the places can offer | The consultation did not cover this section. |
| *Genetic testing* | Explained whether it can be determined if the baby has the disease before it is born, how this might occur and what the risks are. | Explained whether it can be determined if the baby has the disease before it is born and briefly states how this will occur but does not discuss the risks. | States whether it can be determined if the baby has the disease before it is born but does not explain how this might occur or whether there are any risks. | The consultation did not cover this section. |
| *Communication skills and use of genetic terminology* | The information was very well communicated and easy to understand and genetic terminology was used where appropriate. | The information was mostly well communicated and easy to understand and genetic terminology was used where appropriate. | The information was not well communicated and hard to understand. Genetic terminology was not used correctly **OR** the group gave the clients their support material and did not offer any further explanations. | The consultation did not cover this section. |
| *Use of support material* | The group supported their explanation with other material such as audio or visual components. These components were relevant, creative and easy to understand. | The group supported their explanation with other material such as audio or visual components. These components were relevant and mostly easy to understand. | The group supported their explanation with other material such as audio or visual components but it was difficult to see their relevance or understand the content. | The consultation did not cover this section. |
| *Group Work* | All group members contributed equally during the consultation | The group members mostly contributed equally during the consultation. | One group member was mostly responsible for the consultation and received minimal assistance. | One group member completed the consultation and received no assistance. |
| *Counselling Skills* | The group were extremely sensitive to the emotions of their clients and delivered distressing information in a respectful and compassionate manner. | The group were mostly sensitive to the emotions of their clients and delivered distressing information in a respectful and compassionate manner. | The group needed to be more sensitive to the emotions of their clients and delivered distressing information in a respectful and compassionate manner. | The group showed no sensitivity to the emotions of their clients and did not deliver distressing information in a respectful and compassionate manner. |

**State what chances your child have of inheriting the disease, according to the counsellors:** \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Comments:** \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Your Guide to Being a Good Genetic Counsellor**

* Act as if the session was occurring in real life – greet your clients, ask them how they are and what has prompted them to make an appointment with you.
* Ensure everyone in your group contributes to the counselling session.
* Use easy to understand terms.
* Don’t talk at your clients – this is not a presentation, it is a conversation.
* Ensure you are sensitive to the emotions of your clients. The information you are providing them may be distressing.
* Don’t tell them whether or not they should have children – provide them with the information they require to make an informed decision.
* Ask them questions to gauge their understanding.

**Your Guide to Being a Good Client**

* Introduce yourself to the counsellors and tell them about your circumstances (which are stated on the scenario)
* Ensure you ask questions if you do not understand what the client is telling you. For example:
  + Can you explain that further?
  + Where can I find more information on that?
* Ask questions to prompt your counsellor about the information you, as potential parents, are interested in. For example:
  + What are the chances that my child will inherit this disease?
  + How will my child be affected by the disease? Will they still lead a normal life? How long with they live?
  + What kind of symptoms will they experience?
  + Is there any treatment for the disease?
  + Is the disease curable?
  + What support will I have to provide my child? (e.g. at home, at school, hospital visits)
  + Can my child be tested for the disease before birth?
  + How is this likely to affect my life?
  + Where can we get more information?
* At the end of the consultation thank the counsellors for their time



Genetic counselling Research notes

Bibliography must be listed here